

# Coriell Cell Repositories and the Sample Submission Process

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# Outline of the Presentation

- Brief overview of the Coriell Cell Repositories
- Diseases for which characterized mutations are available
- Needs analysis
- Diseases currently being solicited: submission process and requirements
- Considerations for the NIGMS Repository

# Coriell Institute for Medical Research



# Coriell Institute for Medical Research

- Private, Not-for-profit Research Institute
- Research programs in
  - Genetics
  - Cancer
  - Stem Cell Biology
  - Human Variation
  - Physical Anthropology
  - Bioinformatics
- Coriell Cell Repositories
  - Biomaterials collection, storage and distribution
  - Biomaterials research

# Coriell Cell Repositories

Mission: To develop high quality reagents for genetic research and to provide these to qualified investigators as broadly and openly as possible



# Sources of Funding for CCR Activities

- National Institutes of Health
- National Science Foundation
- Centers for Disease Control and Prevention
- Department of Defense
- Pharmaceuticals
- Private Disease Foundations

# Coriell Cell Repositories

## Major Collections

- National Institute for General Medical Sciences (NIGMS) Human Genetic Cell Repository
- National Institute on Aging (NIA) Aging Cell Repository
- National Institute of Neurological Disorders and Stroke (NINDS) Human Genetics Resource Center
- National Cancer Institute (NCI) Cooperative Family Registry for Breast Cancer Studies
- National Cancer Institute (NCI) Cooperative Family Registry for Colon Cancer Studies
- National Eye Institute (NEI) Age Related Eye Diseases Study
- American Diabetes Association (ADA) Maturity Onset Diabetes Repository
- HBDI Juvenile Diabetes Foundation International (JDFI) Repository
- National Science Foundation (NSF) Integrated Primate Biomaterials and Information Resource
- Crohn's and Colitis Foundation of America Repository
- Many private collections for biotechnology and pharmaceutical partners

# Collections Openly Available with Characterized Mutations

- NIGMS - has 2,495 cultures from 172 different diseases for which a gene has been cloned: 608 samples from 72 diseases have characterized mutations
- NIA - 49 samples from six diseases
- NINDS - 6 samples representing two genes
- CDC - 27 samples representing 15 mutations in eight genes



# Genes for Which Characterized Mutations are Available

- *CFTR* - 54 unique mutations
- *HD* - 138 different sizes from 15, 15 to 86, 22
- *FMR1* - 29 different sizes
- *BRCA1* - 24 unique mutations
- *FRDA* - 48 samples with expansions
- *GALT* - 7 unique mutations

# Needs Analysis

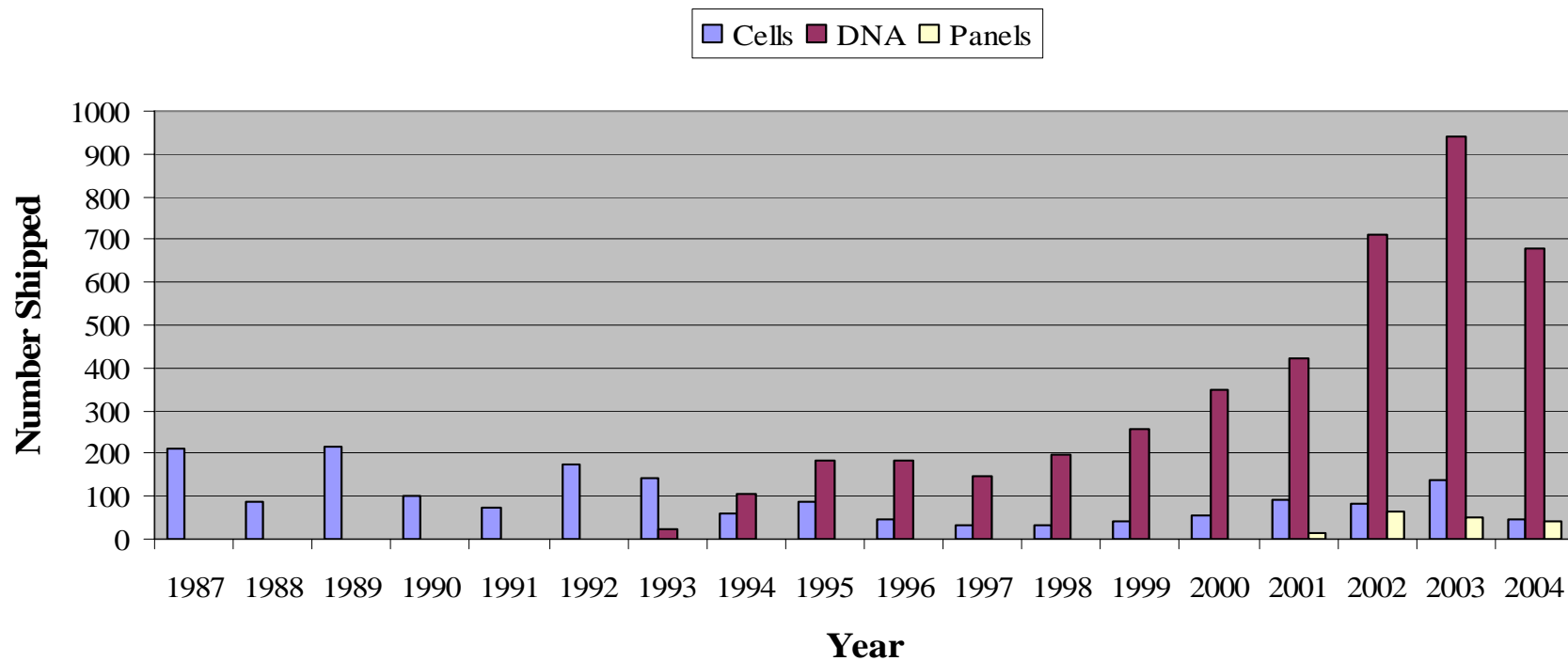
- Unsuccessful searches
- Shipments of biomaterials
- Survey Data

# Unsuccessful Searches

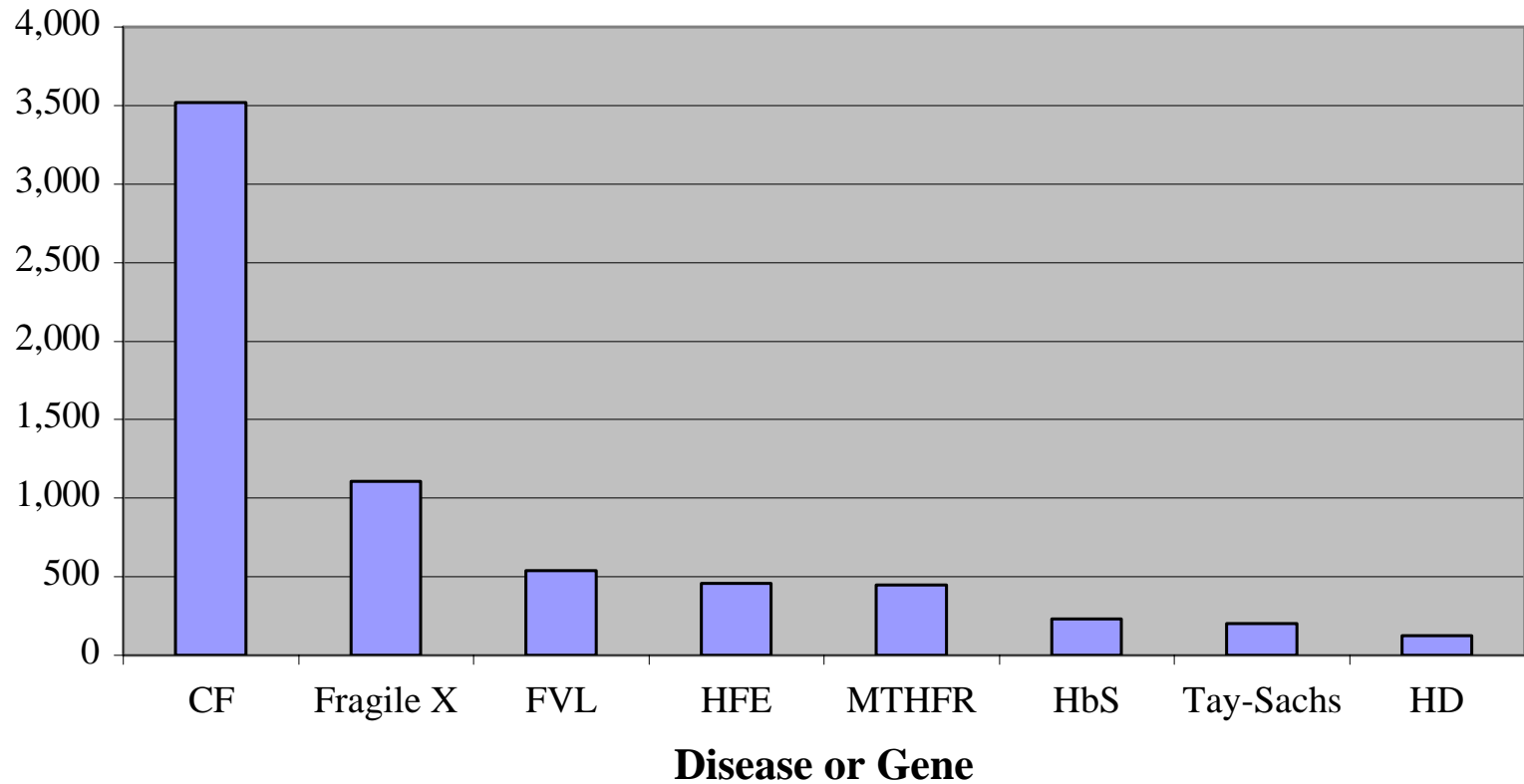
- 26 *CFTR* mutations (I507V, S1251N, A549N, 1898+1G>T, 3199DEL6, R347H, W1089X, Y122X - and 18 others)
- SCA2, SCA8, and SCA 10
- 2 *BRCA1* mutations (3819del4, CYS64GLY)
- Two mitochondrial mutations (A3243G, A1555G)
- One *Cx26* mutation (167delT)
- One *PAH* mutation [R408W (Phenylketonuria)]
- One *ASPA* mutation [E285A(Canavan disease)]
- One *IKBKAP* mutation [R696P(familial dysautonomia)]
- One *GALT* mutations [S135L (galactosemia)]
- 12 others

# Shipments from the NIGMS Repository

## Shipments of Cystic Fibrosis Samples from the NIGMS Repository



## Shipments of Samples since January 1, 2000



# Survey Results

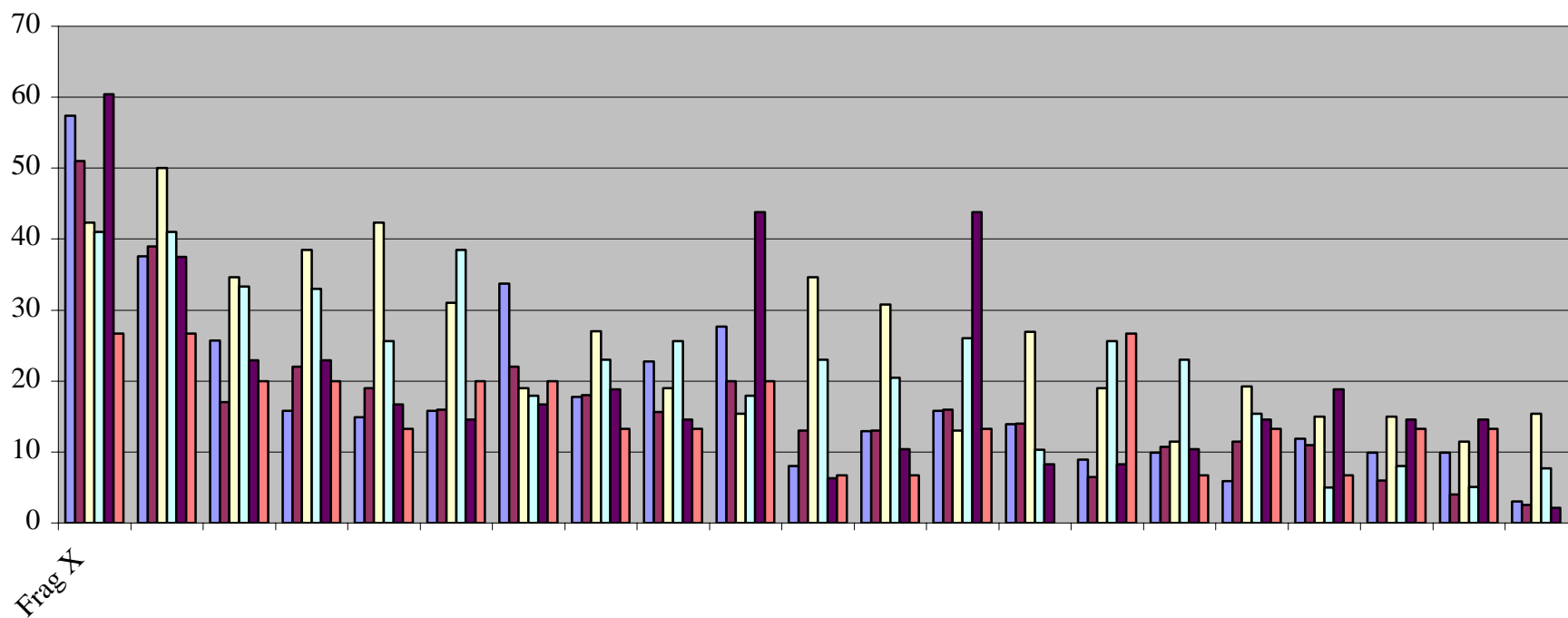
# Survey Results

## Requests for Characterized Mutations

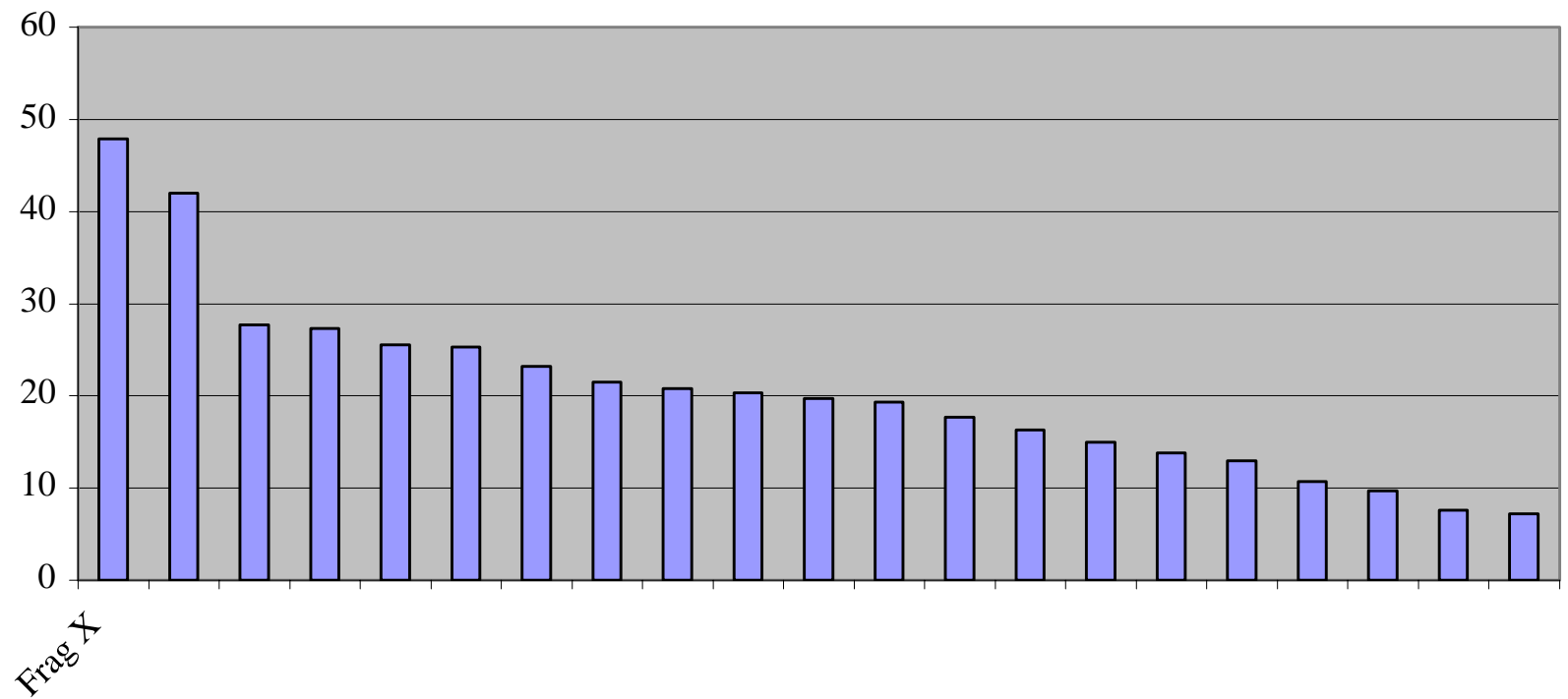
- Survey administered to 603 individuals at six meetings
- 58.4% indicated that they would use samples with characterized mutations
- Respondents were queried about 21 diseases or genes



## Requests for Characterized Mutations from Surveys



## Requests for Characterized Mutations from Surveys

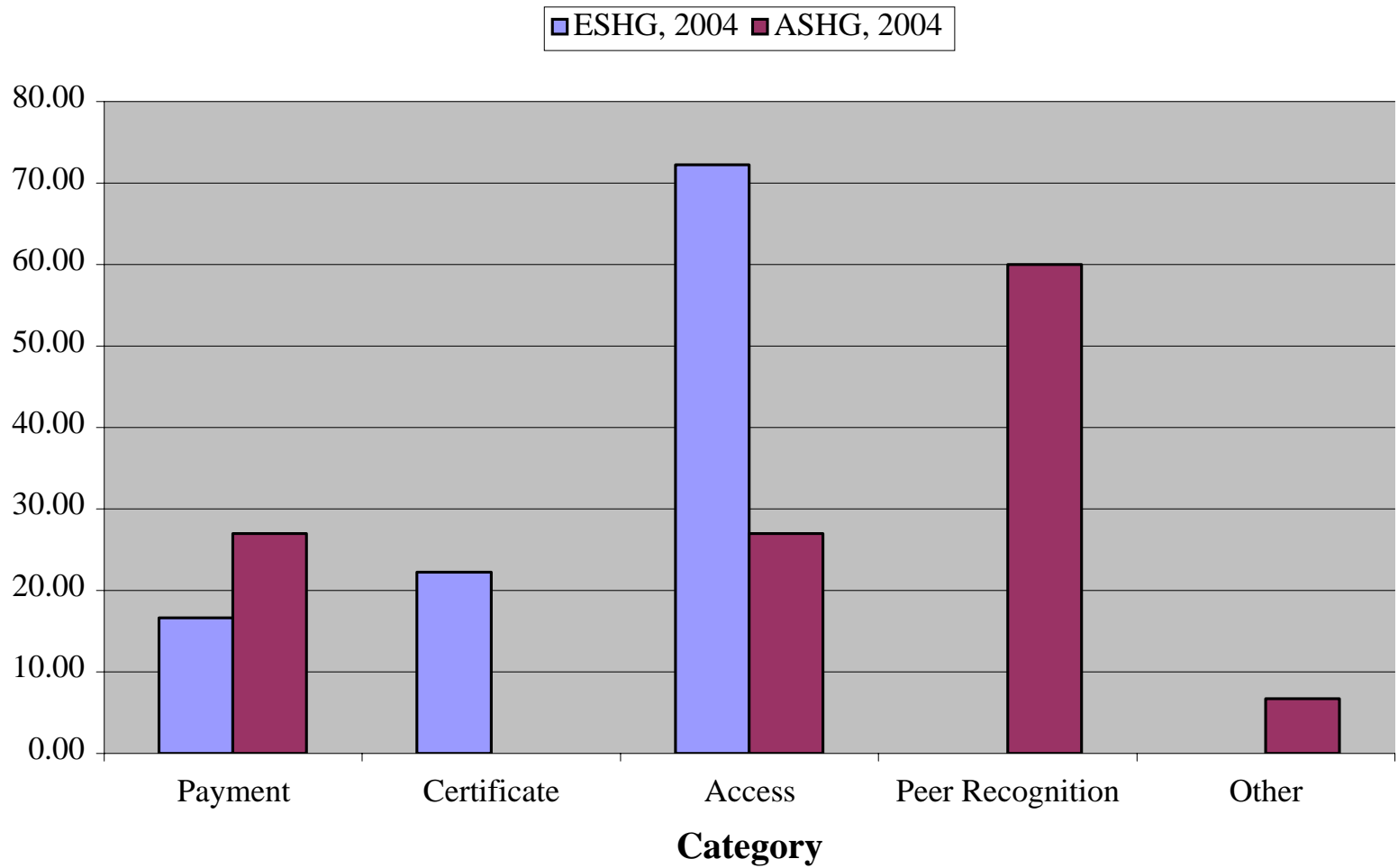


# Survey Results

## Incentives

- Survey administered to 120 individuals at two meetings (ESHG 2004 and ASHG 2004)
- Question posed: What do you see as a fair and reasonable incentive for contributing biomaterials?
  - Payment
  - Certificate of appreciation
  - Access to biomaterials
  - Peer recognition
  - Other

## Results from Surveys about Incentives for Submitters



# Survey Results

## Concerns about Submitting

- Survey administered to 33 individuals at one meeting (ASHG 2004)
- Question posed: What are your concerns for contributing biomaterials?
  - Logistical issues, e.g., informed consent (46%)
  - Not clear about procedures involved (33%)
  - No direct benefit to my work (27%)
  - Takes too much time (7%)
  - Other - please specify: HIPAA

# Submission Process

- For identifiable material:
  - Submission form
    - No personal identifying information is to be included, i.e., none of the 18 specific elements which can identify an individual
  - Unsigned copy of informed consent
- For non-identifiable material (residual blood):
  - Submission form

# Considerations for the NIGMS Repository

- For what diseases would samples be collected?
- How many samples would be collected for each disease?
- Will any phenotypic data be collected for these materials?
- Will these samples be of use to the research community?

# Considerations for the NIGMS Repository, continued

- With regard to distribution, will this work with the current policies for the repository?
- Will the consent used to collect these permit use for research?



# Acknowledgements

- Joe, Bin, Lisa, and Laurina for many discussions
- All of the laboratories who have helped to verify the materials in the NIGMS and the CDC collections

